Complete Summary

GUIDELINE TITLE

Prenatal screening for fetal aneuploidy.

BIBLIOGRAPHIC SOURCE(S)

Summers AM, Langlois S, Wyatt P, Wilson RD, Society of Obstetricians and Gynaecologists of Canada. Prenatal screening for fetal aneuploidy. J Obstet Gynaecol Can 2007 Feb;29(2):146-61. [108 references] PubMed

GUIDELINE STATUS

This is the current release of the guideline.

COMPLETE SUMMARY CONTENT

SCOPE

DISEASE/CONDITION(S)

- Fetal aneuploidy (e.g., Down syndrome and trisomy 18)
- Pregnancy

GUIDELINE CATEGORY

Risk Assessment
Screening

CLINICAL SPECIALTY

Medical Genetics
Obstetrics and Gynecology
Radiology

INTENDED USERS

Advanced Practice Nurses
Patients
Physician Assistants
Physicians

GUIDELINE OBJECTIVE(S)

- To provide recommendations on maternal screening for fetal aneuploidy (e.g., Down syndrome and trisomy 18) in pregnancy
- To develop a Canadian consensus document

TARGET POPULATION

All pregnant women in Canada

INTERVENTIONS AND PRACTICES CONSIDERED

Noninvasive Prenatal Screening for Aneuploidy:

- Offering screening to all pregnant women, regardless of age
- Maternal age combined with first trimester screening: nuchal translucency (NT) maternal serum biochemical markers (pregnancy-associated plasma protein-A [PAPP-A] and free beta-human chorionic gonadotrophin [beta-hCG])
- Maternal age combined with second trimester screening: maternal serum alpha fetoprotein (MSAFP) with two other maternal serum markers

  Combined first and second trimester screening options; integrated prenatal screening and serum integrated prenatal screening, contingent screening, and sequential screening

- Two-step integrated screening, which includes first and second trimester serum screening with or without nuchal translucency (integrated prenatal screening [IPS], Serum IPS, contingent and sequential)

Note: Invasive prenatal diagnosis would be limited to women who screen above a set risk cut-off level on non-invasive screening or pregnant women who will be 40 years at time of delivery who, after counseling, chose to go directly to amniocentesis/chorionic villi sampling.

MAJOR OUTCOMES CONSIDERED

Performance of screening options in relation to:

- Detection rate or sensitivity
- False-positive rate
- Positive rate
- Positive predictive value

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

Searches of Electronic Databases

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

A MEDLINE search was carried out to identify papers related to this topic that were published between 1982 and 2006. Practices across Canada were surveyed.

NUMBER OF SOURCE DOCUMENTS

Not stated

METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE

Weighting According to a Rating Scheme (Scheme Given)

RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE

Level of Evidence*

I: Evidence obtained from at least one properly designed randomized controlled trial.

II-1: Evidence from well-designed controlled trials without randomization.

II-2: Evidence from well-designed cohort (prospective or retrospective) or case–control studies, preferably from more than one centre or research group.

II-3: Evidence from comparisons between times or places with or without the intervention. Dramatic results from uncontrolled experiments (such as the results of treatment with penicillin in the 1940s) could also be included in this category.

III: Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees.

*Adapted from the Evaluation of Evidence criteria described in the Canadian Task Force on the Periodic Health Exam.

METHODS USED TO ANALYZE THE EVIDENCE

Systematic Review

DESCRIPTION OF THE METHODS USED TO ANALYZE THE EVIDENCE

Not stated

METHODS USED TO FORMULATE THE RECOMMENDATIONS

Expert Consensus

DESCRIPTION OF METHODS USED TO FORMULATE THE RECOMMENDATIONS

Not stated

RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS

Classification of Recommendations*

A. There is good evidence to recommend the clinical preventive action.

B. There is fair evidence to recommendation the clinical preventive action.

C. The existing evidence is conflicting and does not allow making a recommendation for or against use of the clinical preventive action; however, other factors may influence decision-making.

D. There is fair evidence to recommend against the clinical preventive action.

E. There is good evidence to recommend against the clinical preventive action.

I. There is insufficient evidence (in quantity or quality) to make a recommendation; however, other factors may influence decision-making.

*Adapted from the Classification of Recommendations criteria described in the Canadian Task Force on the Periodic Preventive Health Exam Care.

COST ANALYSIS

A detailed cost-benefit analysis of the implementation of this guideline has not been done, since this would require health surveillance and research and health resources not presently available; however, these factors need to be evaluated in a prospective approach by provincial and territorial initiatives.

METHOD OF GUIDELINE VALIDATION

Internal Peer Review

DESCRIPTION OF METHOD OF GUIDELINE VALIDATION

This guideline has been reviewed and approved by the Executive and Council of the Society of Obstetricians and Gynecologists of Canada and by the Board of the Canadian College of Medical Geneticists.

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

The level of evidence (I-III) and classification of recommendations (A-E, I) are defined at the end of the "Major Recommendations" field.

Changing the Standard of Care: Screening by Maternal Age Only Limited to Women Aged 40 or Over at Estimated Time of Delivery

1. All pregnant women in Canada, regardless of age, should be offered through an informed consent process a prenatal screening test for the most common clinically significant fetal aneuploidies in addition to a second trimester ultrasound for dating, growth and anomalies. **(I-A)**
2. Maternal age screening is a poor minimum standard for prenatal screening for aneuploidy and should be removed as an indication for invasive testing. Amniocentesis/chorionic villi sampling (CVS) should not be provided without multiple marker screening results except for women over the age of 40. Patients should be counseled accordingly. **(I-A)**

Choosing a Screen

3. In 2007, as a minimum standard, any prenatal screen offered to Canadian women should have a 75% detection rate with no more than a 5% false positive rate for Down syndrome. The performance of the screen should be substantiated by annual audit. **(III-B)**

Review of Screening Options

First Trimester Screening: Nuchal Translucency (NT) Combined with Biochemical Markers

4. First trimester nuchal translucency should be interpreted for risk assessment only when performed by sonographers/sonologists trained and accredited to provide this service and with ongoing quality assurance. **(II-2A)** It should not be offered as a screen without biochemical markers except in the context of multiple gestation pregnancies. **(I-A)**
5. For women who undertake first trimester screening (FTS), second trimester serum alpha fetoprotein (AFP) screening and/or ultrasound examination is recommended to screen for open neural tube defect (ONTD). **(II-1A)**

Combined First and Second Trimester Options

Serum Integrated Prenatal Screening

6. First trimester screening (FTS), the first step of integrated screening (with or without nuchal translucency), contingent, and sequential screening are performed in an early and relatively narrow time window. Timely referral is critical to ensure women are able to undergo the type of screening test they have chosen. **(II-1A)**

7. Soft markers or anomalies in the 18- to 20-week ultrasound can be used to modify the a priori risk of aneuploidy established by age or prior screening provided the scan is undertaken in an established centre performing tertiary level ultrasound. In the absence of ultrasound soft markers or anomalies, a negative likelihood ratio of 0.5 should be used. **(II-2B)**. Evaluation of the fetal nasal bone in the first trimester remains technically difficult and should not be incorporated as a screen until locally established as an effective risk assessment tool. **(III-D)**

General Considerations

8. Health care providers should be aware of the screening modalities available in their province or territory. **(III-B)**
9. Screening programs should be implemented with resources that support audited screening and diagnostic laboratory services, ultrasound, genetic counselling services, patient and health care provider education, and high quality diagnostic testing, as well as resources for administration, annual clinical audit, and data management. In addition, there must be the flexibility and funding to adjust the program to new technology and protocols. **(II-3B)**
10. Screening programs should show respect for the needs and quality of life of persons with disabilities. Counselling should be nondirective and should respect a woman's choice to accept or to refuse any or all of the testing or options offered at any point in the process. **(III-I)**
11. By 2008, screening programs should aim to provide a screen that, as a minimum, offers women who present in first trimester a detection rate of 75% for Down syndrome, with no more than a 3% false positive rate. **(III-B)**

Definitions:

Level of Evidence*

I: Evidence obtained from at least one properly designed randomized controlled trial.

II-1: Evidence from well-designed controlled trials without randomization.

II-2: Evidence from well-designed cohort (prospective or retrospective) or case–control analytic studies, preferably from more than one centre or research group.

II-3: Evidence from comparisons between times or places with or without the intervention. Dramatic results in uncontrolled experiments (such as the results of treatment with penicillin in the 1940s) could also be included in this category.

III: Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees.

Classification of Recommendations**

A. There is good evidence to recommend the clinical preventive action.

B. There is fair evidence to recommendation the clinical preventive action.

C. The existing evidence is conflicting and does not allow making a recommendation for or against use of the clinical preventive action; however, other factors may influence decision-making.

D. There is fair evidence to recommend against the clinical preventive action.

E. There is good evidence to recommend against the clinical preventive action.

I. There is insufficient evidence (in quantity or quality) to make a recommendation; however, other factors may influence decision-making.

*The quality of evidence reported in these guidelines has been adapted from the Evaluation of Evidence criteria described in the Canadian Task Force on the Periodic Health Exam.

**Recommendations included in these guidelines have been adapted from the Classification of Recommendations criteria described in the Canadian Task Force on the Periodic Health Exam.

CLINICAL ALGORITHM(S)

None provided

EVIDENCE SUPPORTING THE RECOMMENDATIONS

TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS

The type of supporting evidence is identified and graded for each recommendation (see "Major Recommendations").

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

POTENTIAL BENEFITS

These guidelines are intended to reduce the number of amniocenteses done when maternal age is the only indication. This will have the benefit of reducing the numbers of normal pregnancies lost because of complications of invasive procedures.

POTENTIAL HARMS

Any screening test has an inherent false positive rate, which may result in undue anxiety.

QUALIFYING STATEMENTS

QUALIFYING STATEMENTS

This guideline reflects emerging clinical and scientific advances as of the date issued and is subject to change. The information should not be construed as dictating an exclusive course of treatment or procedure to be followed. Local institutions can dictate amendments to these opinions. They should be well documented if modified at the local level. None of these contents may be reproduced in any form without prior written permission of the Society of Obstetricians and Gynaecologists of Canada (SOGC).

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES

IOM CARE NEED

Staying Healthy

IOM DOMAIN

Effectiveness
Patient-centeredness
Safety

IDENTIFYING INFORMATION AND AVAILABILITY

BIBLIOGRAPHIC SOURCE(S)

Summers AM, Langlois S, Wyatt P, Wilson RD, Society of Obstetricians and Gynaecologists of Canada. Prenatal screening for fetal aneuploidy. J Obstet Gynaecol Can 2007 Feb;29(2):146-61. [108 references] PubMed

ADAPTATION

Not applicable: The guideline was not adapted from another source.

DATE RELEASED

2007 Feb

GUIDELINE DEVELOPER(S)

Canadian College of Medical Geneticists - Professional Association
Society of Obstetricians and Gynaecologists of Canada - Medical Specialty Society

SOURCE(S) OF FUNDING

Society of Obstetricians and Gynaecologists of Canada

GUIDELINE COMMITTEE

Society of Obstetricians and Gynaecologists of Canada Genetics Committee

Canadian College of Medical Geneticists Committee on Prenatal Diagnosis

Society of Obstetricians and Gynaecologists of Canada Diagnostic Imaging Committee

COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

Principal Authors: Anne M. Summers, MD, North York ON; Sylvie Langlois MD, Vancouver BC; Phil Wyatt, MD, PhD, Toronto ON; R. Douglas Wilson, MD, Philadelphia PA

SOGC Genetics Committee Members: R. Douglas Wilson (*Chair*), MD, Philadelphia PA; Victoria Allen, MD, Halifax NS; Claire Blight, RN, Halifax NS; Valerie Desilets, MD, Montreal QC; Alain Gagnon MD, Vancouver BC; Jo-Ann Johnson, MD, Calgary AB; Sylvie Langlois, MD, Vancouver BC; Anne M. Summers, MD, North York ON; Phil Wyatt, MD, PhD, Toronto ON

CCMG Committee on Prenatal Diagnosis Members: Sylvie Langlois (*Chair*), MD, Vancouver BC; David Chitayat, MD, Toronto ON; Albert E. Chudley, MD, Winnipeg MB; Sandra A. Farrell, MD, Mississauga ON; Michael T. Geraghty, MD, Ottawa ON; Chumei Li, MD, PhD, Toronto ON; Sarah M. Nikkel, MD, Ottawa ON; Andrea Shugar, MS, Toronto ON; Anne Summers, MD, Toronto ON; Frederique Tihy, PhD, Montreal QC

SOGC Diagnostic Imaging Committee Members: Lucie Morin (*Chair*), MD, Montreal QC; Michael Van den Hof (*Past Chair*), MD, Halifax NS; Stephen Bly, MD, Ottawa ON; Robert Gagnon, MD, London ON; Barbara Lewthwaite, RN, Winnipeg MB; Yvonne M. Cargill, MD, Ottawa ON; Kenneth Ian Lim, MD, Vancouver BC; Annie Ouellet, MD, Sherbrooke QC

FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

Not stated

GUIDELINE STATUS

This is the current release of the guideline.

GUIDELINE AVAILABILITY

Electronic copies: Available in Portable Document Format (PDF) from the Society of Obstetricians and Gynaecologists of Canada Web site.

Print copies: Available from the Society of Obstetricians and Gynaecologists of Canada, La société des obstétriciens et gynécologues du Canada (SOGC) 780 promenade Echo Drive Ottawa, ON K1S 5R7 (Canada); Phone: 1-800-561-2416

AVAILABILITY OF COMPANION DOCUMENTS

A listing of screening centres and clinics in Canada is provided in the Appendix C of the original guideline document.

PATIENT RESOURCES

None available

NGC STATUS

This NGC summary was completed by ECRI Institute on February 5, 2009. The information was verified by the guideline developer on March 4, 2009.

COPYRIGHT STATEMENT

DISCLAIMER

NGC DISCLAIMER

Date Modified: 3/30/2009